PRICKLE1 gene

prickle planar cell polarity protein 1

Normal Function

The *PRICKLE1* gene provides instructions for making a protein called prickle homolog 1. The function of this protein is unclear, although it appears to play an important role in the development of the nervous system. Prickle homolog 1 is likely part of a chemical signaling pathway known as noncanonical Wnt signaling. During development before birth, noncanonical Wnt signaling helps to determine the position of various components within cells (cell polarity). This pathway also regulates the movement of nerve cells (neurons) in the developing nervous system.

Studies suggest that prickle homolog 1 interacts with other proteins, including RE1-silencing transcription factor (REST). The REST protein regulates several critical genes in neurons by turning off (suppressing) their activity. To regulate these genes, REST must enter the nucleus and attach (bind) to particular regions of DNA. Researchers believe that prickle homolog 1 controls REST by transporting it out of the nucleus, which prevents it from binding to DNA and suppressing gene activity. It remains unclear how the interaction between prickle homolog 1 and REST contributes to the normal development of the nervous system.

Health Conditions Related to Genetic Changes

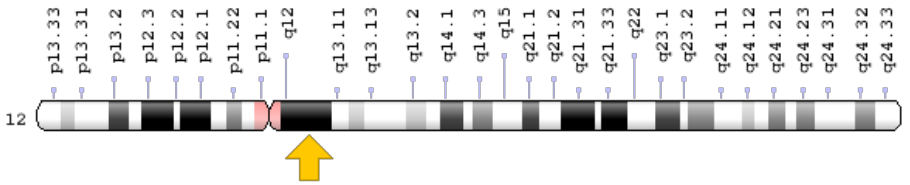
PRICKLE1-related progressive myoclonus epilepsy with ataxia

At least three mutations in the *PRICKLE1* gene have been identified in people with *PRICKLE1*-related progressive myoclonus epilepsy with ataxia. Each mutation changes a single protein building block (amino acid) in the prickle homolog 1 protein. One of the known mutations appears to disrupt the interaction between prickle homolog 1 and REST, blocking the transport of REST out of the nucleus. As a result, REST may inappropriately suppress certain genes in the developing nervous system. It is unclear how mutations in the *PRICKLE1* gene lead to movement problems, seizures, and the other features of *PRICKLE1*-related progressive myoclonus epilepsy with ataxia.

Chromosomal Location

Cytogenetic Location: 12q12, which is the long (q) arm of chromosome 12 at position 12

Molecular Location: base pairs 42,458,338 to 42,589,770 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- EPM1B
- FLJ31627
- FLJ31937
- MGC138902
- MGC138903
- PRIC1_HUMAN
- prickle homolog 1
- prickle-like 1
- REST (RE-1 silencing transcription factor)/NRSF (neuron-restrictive silencer factor)-interacting LIM domain protein
- REST/NRSF-interacting LIM domain protein
- RILP

Additional Information & Resources

Educational Resources

- The WNT Homepage, Stanford University https://web.stanford.edu/group/nusselab/cgi-bin/wnt/

GeneReviews

- PRICKLE1-Related Progressive Myoclonus Epilepsy with Ataxia
  https://www.ncbi.nlm.nih.gov/books/NBK9674

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28PRICKLE1%5BTIAB%5D%29+OR+%28RILP%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- PRICKLE, DROSOPHILA, HOMOLOG OF, 1
  http://omim.org/entry/608500

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PRICKLE1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PRICKLE1%5Bgene%5D
- HGNC Gene Family: LIM domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/1218
- HGNC Gene Family: Prickle planar cell polarity proteins
  http://www.genenames.org/cgi-bin/genefamilies/set/1184
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=17019
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/144165
- UniProt
  http://www.uniprot.org/uniprot/Q96MT3

Sources for This Summary

- Bassuk AG, Wallace RH, Buhr A, Buller AR, Afawi Z, Shimojo M, Miyata S, Chen S, Gonzalez-Alegre P, Griesbach HL, Wu S, Nashelsky M, Vladar EK, Antic D, Ferguson PJ, Cirak S, Voit T, Scott MP, Axelrod JD, Gurnett C, Daoud AS, Kivity S, Neufeld MY, Mazarib A, Straussberg R, Walid S, Korczyn AD, Slusarski DC, Berkovic SF, El-Shanti HI. A homozygous mutation in human PRICKLE1 causes an autosomal-recessive progressive myoclonus epilepsy-ataxia syndrome. Am J Hum Genet. 2008 Nov;83(5):572-81. doi: 10.1016/j.ajhg.2008.10.003. Epub 2008 Oct 30.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/18976727
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2668041/
- Katoh M, Katoh M. Identification and characterization of human PRICKLE1 and PRICKLE2 genes as well as mouse Prickle1 and Prickle2 genes homologous to Drosophila tissue polarity gene prickle. Int J Mol Med. 2003 Feb;11(2):249-56.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/12525887
- Shimojo M, Hersh LB. Characterization of the REST/NRSF-interacting LIM domain protein (RILP): localization and interaction with REST/NRSF. J Neurochem. 2006 Feb;96(4):1130-8. Epub 2006 Jan 17.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16417580
- Shimojo M, Hersh LB. REST/NRSF-interacting LIM domain protein, a putative nuclear translocation receptor. Mol Cell Biol. 2003 Dec;23(24):9025-31.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/14645515
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC309669/
- Tao H, Manak JR, Sowers L, Mei X, Kiyonari H, Abe T, Dahdaleh NS, Yang T, Wu S, Chen S, Fox MH, Gurnett C, Montine T, Bird T, Shaffer LG, Rosenfeld JA, McConnell J, Madan-Khetarpal S, Berry-Kravis E, Griesbach H, Saneto RP, Scott MP, Antic D, Reed J, Boland R, Ehaideb SN, El-Shanti H, Mahajan VB, Ferguson PJ, Axelrod JD, Lehesjoki AE, Fritzsch B, Slusarski DC, Wemmie J, Ueno N, Bassuk AG. Mutations in prickle orthologs cause seizures in flies, mice, and humans. Am J Hum Genet. 2011 Feb 11;88(2):138-49. doi: 10.1016/j.ajhg.2010.12.012. Epub 2011 Feb 3.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/21276947
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3035715/

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/PRICKLE1

Reviewed: December 2011
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services